



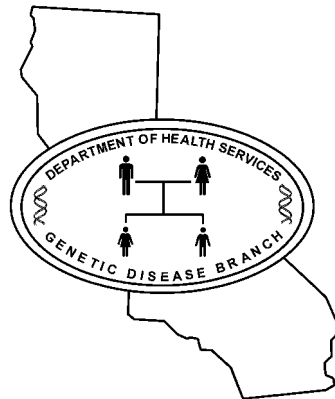
What you should know when your Expanded AFP blood test result is

**Screen Positive: Indicating an increased risk
for neural tube defects or
abdominal wall defects**

POSITIVE: NTD

THE CALIFORNIA EXPANDED AFP SCREENING PROGRAM

the cal ifornia expanded afp screening program
the cal ifornia depart ment of heal th services, genetic disease branch
850 marina bay parkway, f175, richmond, ca 94804
866-718-7915 (tol l free)



As part of your prenatal care you had the Expanded AFP blood test. This screening test helps detect some birth defects such as neural tube defects, abdominal wall defects, Down syndrome and trisomy 18.

This blood test measured three substances in your blood: AFP (alpha-fetoprotein), HCG (human chorionic gonadotropin) and UE (unconjugated estriol). Since there was more than the expected amount of AFP in your blood, your test result was “screen positive”. This indicates an increased risk (chance) that the fetus may have a neural tube defect (such as spina bifida) or an abdominal wall defect.

AFP is a natural substance made by the developing fetus (unborn baby). It is normal for a certain amount of AFP to be in a pregnant woman’s blood. The amount of AFP increases as the fetus grows.

What could have caused your “screen positive” result?

Most of the time, the reason for this result is **NOT** a birth defect. The most common reasons for a high AFP level are:

- you are further along in your pregnancy than was thought, *or*
- you are carrying twins, *or*
- the AFP level in your blood was higher than usual, without any known pregnancy problem.

To help determine *why your result was “screen positive”*, you will be offered diagnostic follow-up services at a State-approved Prenatal Diagnosis Center. There is no additional charge for approved services.

What can diagnostic tests show?

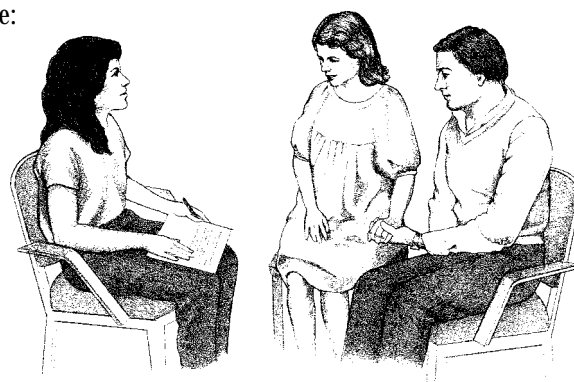
Diagnostic tests can show whether your blood test result was positive because of a change in the due date, or more than one fetus, or a problem with the pregnancy. Diagnostic tests can also show whether the fetus has certain birth defects such as neural tube defects or abdominal wall defects. (See pages 6-7 for information on these birth defects.)

Most women with “screen positive” results have normal follow-up tests and healthy babies.

What are the follow-up services at a State-approved Prenatal Diagnosis Center?

These follow-up services include:

- genetic counseling
- ultrasound
- amniocentesis



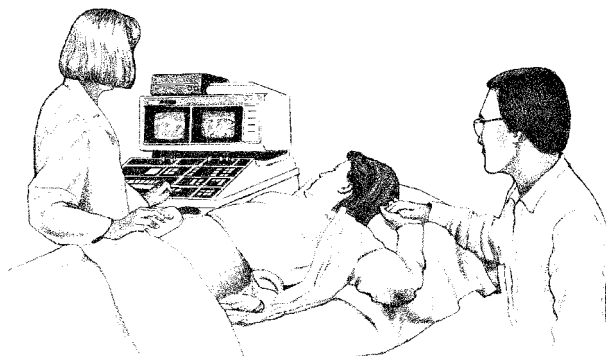
A woman may refuse any of these follow-up services at any time.

Genetic counseling:

A genetic counselor or doctor will discuss your blood test result and what it could mean. Your family's health history will be reviewed. Counseling will give you information to help you make decisions about the follow-up tests. Be sure to talk with the counselor or doctor about any questions or concerns you may have.

Ultrasound:

This test is also called a sonogram. Sound waves are used to make a picture of the fetus. This picture is seen on a special TV screen.



- ***Ultrasound shows how old the fetus really is.***

If the ultrasound shows that the fetus is older than was originally thought, the Expanded AFP blood test result will be recalculated. The new result may actually be “screen negative” for a fetus of that age.

The ultrasound may show that the blood test was done “too late” (after the 20th week of pregnancy).

If the ultrasound dating has made your blood test result “screen negative” or “too late”, it means that the original result was not really positive. In this case, no further follow-up tests are offered by the Program.

- ***Ultrasound shows if you are carrying twins (or triplets).***

If there is more than one fetus, the blood test result may actually be “screen negative” for multiple fetuses. In this case, no further follow-up tests are offered by the Program.

- ***Ultrasound may show whether the fetus has an abnormality.***

If there is a neural tube defect or an abdominal wall defect, it can usually be seen with ultrasound. Ultrasound can also detect some other visible abnormalities.

- ***Ultrasound may show whether there is a pregnancy complication,*** such as a problem with the placenta, bleeding, slow growth, or fetal death.

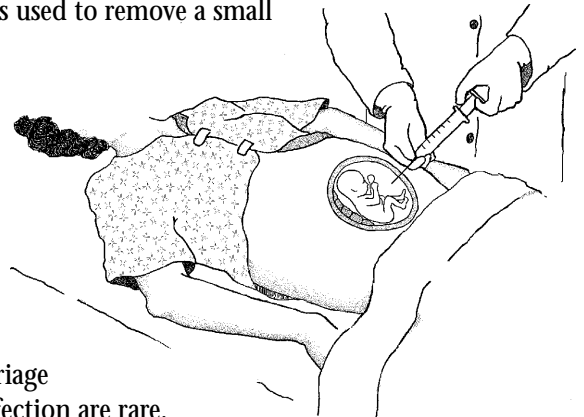
*Amniocentesis is offered • if ultrasound does not explain why there was a high level of AFP,
or
• if ultrasound finds a problem which needs clarification.*

Amniocentesis:

This test involves removing a small amount of the amniotic fluid which surrounds the fetus. First, an ultrasound picture locates the fetus and the fluid. Then a thin needle is used to remove a small amount of the fluid. This procedure sometimes causes brief discomfort.

The amount of AFP in the fluid is measured. The fluid also contains fetal cells. The chromosomes in these cells are examined for abnormalities. Together with ultrasound, amniocentesis will detect nearly all open neural tube defects and abdominal wall defects. Amniocentesis also diagnoses chromosome disorders in the fetus. The results are usually ready in two weeks.

Amniocentesis is considered a safe procedure when performed by medical experts at a State-approved Prenatal Diagnosis Center. The risk of miscarriage following amniocentesis is less than 1%. Problems such as bleeding or infection are rare.



**Sometimes not all of the follow-up tests are needed.
Genetic counseling will explain which tests will be useful.**

Most of the time, the follow-up tests do not show any problems with the fetus. Occasionally, however, a **neural tube defect** or an **abdominal wall defect** is diagnosed. These birth defects are associated with a high AFP level in the mother's blood and in the amniotic fluid.

Neural tube defects:

The neural tube starts at the top of the head and ends at the bottom of the spine. As a fetus is growing, the neural tube becomes the baby's brain and spinal cord. The neural tube is completely formed by 5 weeks after conception. If the neural tube does not develop properly, birth defects can occur.

The two most common neural tube defects are **spina bifida** and **anencephaly**. In the United States, these defects occur in about 1 in every 1,000 births.



Spina bifida:

Infants with spina bifida have an opening in the bones of the spine. When the opening is not covered by skin, it is called **open spina bifida**. With this condition, the nerves that control the lower parts of the body are exposed and damaged. In severe cases the infant may die.

Sometimes the opening is covered by skin, so the nerves are less damaged. This is called **closed spina bifida**. It is less common and will probably not be detected by the Expanded AFP Screening Program.

Most people with open spina bifida have some or all of the following lifelong problems:

- weak or paralyzed lower body
- problems with bladder and bowel control
- bladder and kidney infections
- deformed legs or spine
- “water on the brain” (hydrocephalus)

Some of these problems can be reduced through surgery immediately after birth. Prompt treatment is also necessary to prevent or minimize hydrocephalus, which can cause mental retardation. Medications, physical therapy, and leg braces can help the growing child. Many children and adults with spina bifida lead fulfilling lives.

Anencephaly:

Anencephaly occurs when a large part of the skull is missing and most of the brain does not develop. This defect causes the death of the fetus or newborn.

Abdominal wall defects:

An infant with one of these defects has an abnormal opening on the abdomen. This often causes the intestines and other organs to form outside the body.

An opening on the infant's abdomen where the umbilical cord is attached is called **omphalocele**. This occurs in about 1 out of 4,000 births. An opening beside the umbilical cord is called **gastroschisis**. This occurs in about 1 out of 10,000 births. These openings may be present along with other birth defects.

Surgery soon after birth can often repair an abdominal wall defect.

If the fetus does have one of these birth defects, will the follow-up tests detect it?

Follow-up tests will detect almost all the cases of anencephaly, open spina bifida and abdominal wall defects. These tests should be done at a State-approved Prenatal Diagnosis Center.

What if the follow-up tests show that the fetus has a birth defect?

If a birth defect is found, a doctor or genetic counselor will give the woman or couple information about the defect and how it may affect the fetus. Available treatments and options for continuing or ending the pregnancy will be discussed. If the pregnancy is continued, delivery should take place at a major medical center in order to give the baby the best chances for survival and development.

The Expanded AFP Screening Program does not pay for any other medical services after the follow-up tests. Referrals for further medical and support services are available.

Please remember:

Most women who have had a “screen positive” test result will have normal healthy babies. However, in those pregnancies with a serious birth defect, early detection allows parents to explore their options and make early decisions.

The goal of the Department is to provide high quality, low cost services to all Californians. If you have questions, comments or suggestions about services received through this program please let us know.

Write to:

george c. cunningham, md
the cal ifornia depart ment of heal th services
genetic disease branch
850 marina bay par kway, f175
richmond, ca 94804

